Perspectives

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Getting Ready for the Future: Integration of Genomics into Public Health Research, Policy and Practice in Europe and Globally

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Abstract
The integration of genomics into public health research, policy and practice will be one of the most important future challenges that our health care systems will face. The next decade will provide a window of opportunity to establish infrastructures that will enable the scientific advances to be translated into evidence-based policies and interventions that improve population health. Approaches for national, European and international institutionalization of public health genomics are shown that aim to champion these challenges.

The Challenge

During the past century, achievements in public health have led to enormous improvements and benefits in the health and life expectancy of people around the world. Immunization programs and better sanitation practices resulted in the eradication or decreased incidences of many infectious diseases as well as in safer food and water supplies. Advances in occupational safety considerably decreased the number of work-related injuries, illnesses and deaths. In the past 30 years, identification of behavioral risk factors, such as smoking, inactivity and poor dietary habits, gave rise to educational interventions resulting in a decline in death rates from certain chronic diseases. For future achievements in public health, the Office of Genomics and Disease Prevention, Centers of Disease Control, in Atlanta, Ga., USA, predicts that to continue making significant strides, the effectiveness of public health interventions must be strengthened by more fully incorporating knowledge of internal, host-specific factors and their interactions with environmental exposures including the social environment and life-styles.

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In the past 20 years, the advances in genome research have revolutionized the knowledge of the role of inheritance in health and disease. Nowadays, it is known that the DNA determines not only the cause of single-gene disorders, which affect millions of people worldwide, but also predispositions ('susceptibilities'), which are based on genotype and haplotype variants, to common diseases. The new technologies will allow researchers to examine genetic mutations at the functional genomic unit level, and to better understand the significance of environmental factors such as chemical agents, nutrition or personal behavior in relation to the etiology of diseases like cardiovascular diseases, allergies, cancer, psychiatric disorders or infectious diseases.

Evidently, these rapid advances in genomics and accompanying technologies are triggering a shift in the comprehension of health and disease as well as in the understanding of new approaches to prevention and therapy. Clarifying the general conditions under which genomic knowledge can be put best into practice in the field of public health, paying particular attention to the ethical, legal and social implications, is currently the most pressing task in the emerging field of public health, variously dubbed public health genetics (PHG) or public health genomics (PHG). Aiming to apply genetic and molecular science in the promotion of health and disease prevention through the combined efforts of society, integral to its activities is the dialogue with all stakeholders in society, including industry, governments, health professionals and the public in general. Thus, the integration of genomics into public health research, policy and practice will be one of the most important challenges for our health care systems in the future. Expertise is already feasible and can be clustered and evaluated for a socially accountable use.

Interestingly enough, European and US public health institutions and platforms like the PHG Unit in Cambridge (PHGU), UK, the German Center for PHG in Bielefeld (DZPHG) or the US Office of Genomics and Disease Prevention at the Centers for Disease Control and Prevention in Atlanta, who work closely together with researchers from genetic and molecular science ('modern biology') as well as from population science, humanities and social science, are much more optimistic and clear about the relevance of genomics for public health than others. They all have strong links or are even part of the respective national genome research projects in these countries and are translating genomic knowledge from biotechnology through genetic epidemiology into public health ('translational research'). By using methods like horizon scanning, fact finding and monitoring to identify research trends as early as possible, they are already doing a prospective evidence-based evaluation, i.e. an evaluation that is already carried out in the process of basic research and not just in the (retrospective) process of the implementation of public health strategies and policies, which will always tend to lack behind.

In the realm of social policy-making, there is a need to come up with a clear strategy for assessing and translating novel genomic knowledge and application right in time. Policy-makers have now the opportunity to take action. Precondition for immediate action is strategic planning across health programs, promoting genomics competencies among health professionals, enhancing surveillance and epidemiologic capacity to support evidence-based policy-making, building partnerships and seeking input from stakeholders. Sound and well-reflected genetics policies and programs require a timely and coordinated process for evidence-based policy-making that relies on scientific research and ongoing community consultation. An acceptable and maybe delicate balance between providing strong protection of individual interests and enabling society to benefit from the genomic advancements at the same time must be found.

The sheer volume and complexity of this emerging genomic knowledge, and the speed of technological development, are such that the goals of this enterprise can only be achieved by taking an integrated, interdisciplinary European as well as international approach.

In the following, we shortly introduce two well-established national, a recent European and two current worldwide approaches of institutionalizing PHG that may serve as a paradigm for further efforts to champion the progress in molecular biology for the benefit of the population – without neglecting the individual interests and rights.

Public Health Genetics Unit in Cambridge

The PHGU was established in Cambridge, UK, in 1997 to keep abreast of developments in molecular and clinical genetics, and in their ethical, legal, social and public health implications (www.phgu.org.uk). Its aim is to provide a link between academic research, clinical practice and the development of policy within the NHS for genetics and genetic services, including the implications for the funding, development, personnel, organization and provision of those services. Furthermore, among the tasks of the PHGU is to establish mechanisms for the

dialogue within the NHS between geneticists, physicians, public health and primary care professionals on matters related to genetics, molecular medicine and genetic services. Providing an epidemiological and public health perspective on NHS policy development for genetic and related services, including criteria for evaluating genetic tests and genetic screening programs, also belongs to the main fields of work of the PHGU.

Since 2002 the PHGU has functioned as the core facility of the Cambridge Genetics Knowledge Park, one of six established in England and Wales through funding from the Departments of Health, and Trade and Industry. The Cambridge Genetics Knowledge Park has special expertise in the policy and public health aspects of genetics and has sought to catalyze interdisciplinary work between communities engaged in scientific, clinical and public health research as well as in commercial and ethical, legal and social implications.

As a result of the work of the PHGU in the UK, PHG is beginning to be recognized as a small but important aspect of public health science and practice and to have an important role in the shaping of genetics policy within health and health services.

German Center for Public Health Genomics

The DZPHG is a center of excellence and 'think tank' in the field of PHG operating at the national, European and international level (www.public-health-genetics.org). It is an umbrella institution aiming at the advancement of interdisciplinary translational research through various fields of science and the humanities (biomedicine, biology, philosophy/social ethics and bioethics, social and political sciences, economics, biolaw, epidemiology/biostatistics and public health), interdisciplinary and interinstitutional long-term cooperation and exchange across the boundaries of established academic disciplines as well as between relevant stakeholders in the healthcare system. Integration into the University of Applied Sciences in Bielefeld ensures excellent working conditions and provides ample opportunities for training and higher education in the field of PHG.

Main objectives of the DZPHG are

- translational research on the integration of genomics into public health research, policy and practice through national, European and international activities
- systematic horizon scanning, monitoring and surveillance of challenges and tasks of PHG by assessing health needs, health technology and health impact
- improvement in health care at the national, European and international level by establishing a knowledge base for evidence-based policy-making
- interdisciplinary and interinstitutional research and training
- holding meetings open to the scientific community, policy-makers and the general public.

Current European and international activities of the DZPHG related to PHG include

- associated partnership in the European Union funded project Public Health Genomics European Network (PHGEN)
- steering committee membership in the Genome-Based Research and Population Health International Network (GRAPH Int)
- associated partnership in the international non-profit organization Public Population Project in Genomics (P^3G)
- coordination of the German Task Force PHG at the Center for Interdisciplinary Research in Bielefeld
- coordination of the PHG platform in Bielefeld within the German National Genome Research Network
- coordination of the section PHG within the European Association of Public Health (EUPHA).

Public Health Genomics European Network

To consider genetic determinants as a factor contributing to health and as such as a component for public health is a necessary step to enable good health for all – which is the motto for EU health policy. Thus, genetic determinants have to play an eminent role in a new health strategy of the EU that subscribes to the aim of getting good health for all.

In its third report on 'Life Science and Biotechnology', the European Commission has committed itself to gain high quality in genetic testing and to increase 'co-operation and exchange of information in order to enhance coherence and disseminate best practice' [1]. Furthermore, in the work plan 2005 of the 'community action in the field of public health' the European Commission calls for an application for a 'networking exercise … to lead to an inventory report on genetic determinants relevant to public health …'.

Thus, recently a Public Health Genomics European Network (PHGEN) has been funded by the EU (www.phgen.nrw.de), which is coordinated by the Institute of Public Health of North Rhine-Westphalia in Bielefeld, Germany. The network exists of representatives from

genetic and public health experts and representatives from competent authorities of all EU member states, applicant countries, the European Free Trade Association and European Economic Area countries.

The aims of PHGEN are to

- develop links with relevant European Commission programs and actions and with national and regional initiatives
- gather and exchange information concerning best practice in order to assess and prepare the development of European Commission policies, strategies and measures in the field of PHG
- contribute to a high level of health protection and improvement in public health
- take into account the need for supporting member states' actions and enhanced cooperation in the EU context, legal obligations and their implementation
- create self-sustainable mechanisms which enable the member states to coordinate their health-related activities in the field of PHG.

Tasks of this European network include

- establishing a network on PHG in Europe, which, in the long run, can serve for the European Commission as an 'early detection unit' for horizon scanning, fact finding, and monitoring of genetic determinants relevant for public health
- listing key experts for PHG in Europe
- writing an 'inventory report' on genetic predispositions/determinants relevant to population health
- writing a 'state-of-the-art report' on PHG issues and priorities in Europe (e.g. initiatives, legal frameworks or evaluation of current national practices in applying genetic testing).

With the help of this network, across Europe, there will be the chance of scientific advances being timely, effectively, efficiently and socially acceptably translated into evidence-based policies and interventions that improve population health.

Genome–Based Research and Population Health

Via the GRAPH Int Network, international collaboration facilitates the responsible and evidence-based integration of genome-based knowledge and technologies into public policy and into services for improving the health of populations (www.humgen.unmontreal.ca/event/dnasampling).

An international expert meeting was convened with funding from the Rockefeller Foundation at their conference center in Bellagio, Italy, in April 2005, to explore the possibility of establishing an international network to promote the objectives of PHG, to share knowledge and resources, and to ensure equitable access to the benefits of genome-based knowledge by all, including those in developing countries. The meeting was attended by a multidisciplinary group of eighteen experts from Canada, France, Germany, the United Kingdom and the United States.

The participants unanimously agreed to establish an international forum for its promotion, to be known as GRAPH Int. The use of the term Int signifies that the collaboration is not only international but also interdisciplinary and integrated.

The objectives of GRAPH Int are to provide an international forum for dialogue and collaboration, promote relevant research, support the development of an integrated knowledge base, promote education and training, encourage communication and engagement with the public and other stakeholders and inform on public policy.

The vision and the ultimate goal of both the enterprise and the network is the effective translation of genome-based knowledge for the benefit of population health.

Public Population Project in Genomics

P^3G is an international, non-profit organization for the development and management of a multidisciplinary infrastructure in the field of population genomics (www.p3gconsortium.org).

Dedicated to foster collaboration between researchers and projects in the field of population genomics, P^3G develops, in an open and transparent manner, research tools for effective collaboration between biobanks to enable the international research community to share expertise and resources and facilitate knowledge transfer for the health of populations.

The aims of P^3G are to

- create a network in population genomics that will comprise over 3 million participants for epidemiological studies
- provide statistical power for the analysis of complex genetic and environmental determinants of health and disease
- leverage the combined expertise of hundreds of researchers around the world
- promote communication among national and international organizations
- increase the ability to share and generate new knowledge dedicated to improve public health and welfare.

Major issues need to be addressed prior to transferring new genetic discoveries to health care systems. These issues range from the essential step of validating initial findings in other populations to understanding genetics in relation to diseases. Other issues, such as the impact of the environment, the cost implications of genetic testing in relation to benefits for populations and their governments, and the establishment of appropriate guidelines to inform health ministries, health care workers, patients and their relatives also need to be addressed.

Conclusion

The next decade will provide a window of opportunity to establish infrastructures, across Europe and globally, that will enable the scientific advances to be effectively and efficiently translated into evidence-based policies and interventions that improve public health. Policy-makers now have the opportunity to protect consumers, to monitor the implications of genomics for health services, and to assure that genetic advances will be taped to prevent disease and improve health. We now have the chance to prepare public health professionals, the public and policy-makers for the changes to come. The above-presented examples show approaches for national, European and international institutionalization of PHG that serve the aim to champion these challenges.

Reference

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